

CLAIMS

What is claimed is:

5 1. A method of testing a subject to determine if the subject has a predisposition for developing breast cancer which comprises the steps of:

 (a) detecting a mutation in the open reading frame of the ATM gene (SEQ.ID.NO: 1) in a cDNA sample from the subject, which mutation is selected
10 from the group consisting of the mutations set forth in Table 4 and Table 5; or

 (b) detecting a mutation corresponding to a mutation in the open reading frame of the ATM gene (SEQ.ID.NO: 1) in a genomic DNA sample from the subject, which mutation is selected from the group consisting of the
15 mutations set forth in Table 4 and Table 5,

wherein the presence of such mutation indicates that the subject has a predisposition for developing breast cancer.

20 2. The method according to claim 1, wherein said detecting step includes detecting DNA which is characterized by including at least one mutation selected from the group consisting of mutations in position 3161 C->G, position 2572 T->C, position 6235 G->A, position 3118 A->G, position 378 T->A, position 2614 C->T, position 146 C->G, and position 1636 C->G.

3. The method according to claim 1, wherein said detecting step includes detecting DNA which is characterized by including at least two mutations selected from the group consisting of a double mutation in position 3161 (C>G) and position 2572(T>C); and a double mutation in position 6253(G>A) and position 378 (T>A).

4. A method of testing a subject, who has already developed primary breast cancer, to determine if the subject has a predisposition to develop bilateral breast cancer which comprises:

(a) detecting a mutation in the open reading frame of the ATM gene (SEQ.ID.NO: 1) in a cDNA sample from the subject a mutation, which mutation is selected from the group consisting of the mutations set forth in Table 4 and Table 5; or

(b) detecting a mutation corresponding to a mutation in the open reading frame of the ATM gene (SEQ.ID.NO: 1) in a genomic DNA sample from the subject, which mutation is selected from the group consisting of the mutations set forth in Table 4 and Table 5,

wherein the presence of such mutation indicates that the subject has a predisposition to develop bilateral breast cancer.

5. The method according to claim 4, wherein said detecting step includes detecting DNA which is characterized by including at least one mutation selected from the group selected from the group consisting essentially of mutations in position 3161 C->G, position 2572 T->C, position 6235 G->A, position 3118 A->G, position 378 T->A, position 2614 C->T, position 146 C->G, and position 1636 C->G.

6. The method according to claim 4, wherein said detecting step includes detecting DNA which is characterized by including at least two mutations selected from the group consisting of double mutation in position 3161 (C>G) and position 2572(T>C); and double mutation in position 6253(G>A) and position 378 (T>A).

7. An isolated cDNA having a nucleotide sequence which differs from the sequence set forth in SEQ.ID.NO: 1 by a mutation selected from the group consisting of mutations in position 378 T->A, position 3383 A->G, position 1636 C->G, position 2614 C->T, position 6437 G->C, position 2932 T->C, position 2289 T->A, position 6096 A-> T, position 6176 C->T, position 6919 C->T, , position 2442 C->A, position 3925 G->A, position 6067 G->A, position 2119 T->C, position 1810 C->T, and position 4388 T->G.

8. A marker for determining a predisposition for breast cancer, wherein said marker includes a mutation in the open reading frame of the ATM gene (SEQ.ID.NO: 1).

5 9. The marker according to claim 8, wherein said mutation is selected from the group consisting of the mutations set forth in Table 4 and Table 5.

10 10. The marker according to claim 9, wherein said mutation is selected from the group consisting of mutations in position 378 T->A, position 3383 A->G, position 636 C->G, position 2614 C->T, position 6437 G->C, position 2932 T->C, position 2289 T->A, position 6096 A-> T, position 6176 C->T, position 6919 C->T, position 3925 G->A, position 6067 G->A, position 2119 T->C, position 1810 C->T, and position 4388 T->G.